

# What Is Osteogenesis Imperfecta?

## *Fast Facts: An Easy-to-Read Series of Publications for the Public*

Osteogenesis imperfecta (OI) is a disease that causes weak bones that break easily. It is known as brittle bone disease. Sometimes the bones break for no known reason. OI can also cause many other problems such as weak muscles, brittle teeth, and hearing loss. About 20,000 to 50,000 people in the United States have OI.

### **What Causes Osteogenesis Imperfecta?**

OI is caused by a defect in a gene. This happens in one of the genes that control how the body makes type 1 collagen, which helps make bones strong. Most children with OI inherit the faulty gene from a parent. Sometimes the gene that causes OI does not come from a parent. Instead, a change (mutation) in the gene occurs by chance. This happens soon after the child is conceived.

National Institutes of Health  
Osteoporosis and Related Bone  
Diseases~National Resource Center  
2 AMS Circle  
Bethesda, MD 20892-3676

Phone: 202-223-0344;  
1-800-624-BONE (free of charge)  
TTY: 202-466-4315  
Fax: 202-293-2356  
E-mail: [NIAMSBoneInfo@mail.nih.gov](mailto:NIAMSBoneInfo@mail.nih.gov)  
[www.niams.nih.gov/bone](http://www.niams.nih.gov/bone)

The NIH Osteoporosis and Related Bone Diseases~National Resource Center is supported by the National Institute of Arthritis and Musculoskeletal and Skin Diseases with contributions from the National Institute of Child Health and Human Development, National Institute of Dental and Craniofacial Research, National Institute of Diabetes and Digestive and Kidney Diseases, NIH Office of Research on Women's Health, DHHS Office on Women's Health, and the National Institute on Aging.

The National Institutes of Health (NIH) is a component of the U.S. Department of Health and Human Services.



### **What Are the Symptoms of Osteogenesis Imperfecta?**

All people with osteogenesis imperfecta have brittle bones. OI can range from mild to severe and symptoms vary from person to person. Some of the symptoms that people with OI may have are:

- Deformed bones
- Short, small body
- Loose joints
- Muscle weakness
- Sclera (white of the eye) that look blue, purple, or gray
- Triangular face
- Barrel-shaped rib cage
- Curved spine
- Brittle teeth
- Hearing loss (often starting in 20s or 30s)
- Breathing problems
- Type 1 collagen that does not work well
- Not enough collagen.

### **What Are Some Types of Osteogenesis Imperfecta?**

There are four main types of osteogenesis imperfecta.

#### **Type I OI**

Type I osteogenesis imperfecta is the most common and the mildest form.

## What Is Osteogenesis Imperfecta?

*Fast Facts: An Easy-to-Read Series of Publications for the Public*

People with Type I OI are often average height for their family and do not tend to have deformed bones. Their bones may break easily and they may have loose joints and weak muscles.

### Type II OI

Type II osteogenesis imperfecta is the most severe form. Babies with Type II OI usually die from breathing problems at birth or during the first few months.

### Type III OI

People with Type III osteogenesis imperfecta have bones that break easily. Babies are sometimes born with broken bones. X rays may show bones that broke and healed before birth. Adult height of people with this type of OI is less than 4 feet. People with type III OI tend to have barrel chests and breathing problems.

### Type IV OI

Type IV osteogenesis imperfecta is also called moderate OI. People with this type of OI are shorter than average for their age and may have bowing in the long bones of the legs.

### Other Types of OI

Recently, two new types of OI have been described: Types V and VI OI. Except for certain changes in bone, people with these types of OI look like people with the moderate form of OI. A change in the type 1 collagen gene has not been linked to these types of osteogenesis imperfecta.

## How Is Osteogenesis Imperfecta Diagnosed?

No single test can identify osteogenesis imperfecta. To diagnose OI, doctors look at:

- Family history
- Medical history
- Results from a physical exam
- X rays.

Your doctor may also test your collagen (from skin) or genes (from blood). It may take a few weeks to learn the results of the tests. These tests spot OI in 9 out of 10 people who have it.

## How Is Osteogenesis Imperfecta Treated?

Although there is no cure for OI, symptoms can be managed. Treatments for OI may include:

- Care for broken bones
- Care for brittle teeth
- Pain medication
- Physical therapy
- Use of wheelchairs, braces, and other aids
- Surgery.

One type of surgery is called "rodding." Metal rods are put inside the long bones to:

## What Is Osteogenesis Imperfecta?

*Fast Facts: An Easy-to-Read Series of Publications for the Public*

- Strengthen them
- Fix bone deformity
- Prevent bone deformity.

A healthy lifestyle also helps people with OI. You can help prevent broken bones and maintain your health if you:

- Exercise (swimming, water therapy, walking)
- Keep a healthy weight
- Eat a balanced diet
- Do not smoke
- Do not drink a lot of alcohol and caffeine
- Do not take steroid medicines.

Proper care helps children and adults who have OI to:

- Stay active
- Make bones more dense
- Keep muscles strong.

## What Research Is Being Done on Osteogenesis Imperfecta?

No medications are approved for people with OI. But, experts are trying to learn more about:

- The gene that causes OI
- Medications to help people with OI grow (growth hormone)
- Drugs to make bones stronger (bisphosphonates and teriparatide)
- Better devices to use in surgery.

## For More Information About Osteogenesis Imperfecta and Other Related Conditions:

### NIH Osteoporosis and Related Bone Diseases~National Resource Center

2 AMS Circle

Bethesda, MD 20892–3676

Phone: 202–223–0344 or

800–624–BONE (624–2663) (free of charge)

TTY: 202–466–4315

Fax: 202–293–2356

E-mail: NIAMSBoneInfo@mail.nih.gov

[www.niams.nih.gov/bone](http://www.niams.nih.gov/bone)

The information in this publication was summarized in easy-to-read format from a more detailed publication. To view, download, or order the full-text version, visit <http://www.niams.nih.gov/bone>.

## **What Is Osteogenesis Imperfecta?**

*Fast Facts: An Easy-to-Read Series of Publications for the Public*

The NIH Osteoporosis and Related Bone Diseases–National Resource Center acknowledges the assistance of the Osteogenesis Imperfecta Foundation ([www.oif.org](http://www.oif.org)) in the preparation of this publication.

### **For Your Information**

This publication contains information about medications used to treat the health condition discussed here. When this fact sheet was printed, we included the most up-to-date (accurate) information available. Occasionally, new information on medication is released.

For updates and for any questions about any medications you are taking, please contact the U.S. Food and Drug Administration at 1–888–INFO–FDA (1–888–463–6332, a toll-free call) or visit their Web site at [www.fda.gov](http://www.fda.gov).